

Figure 1

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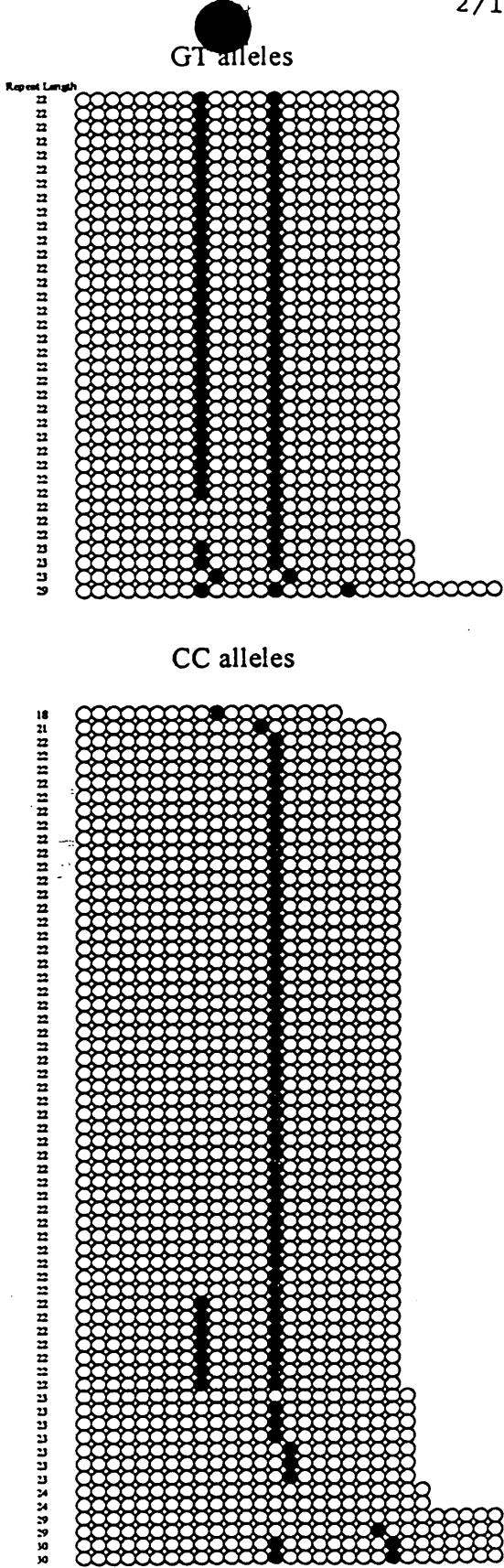
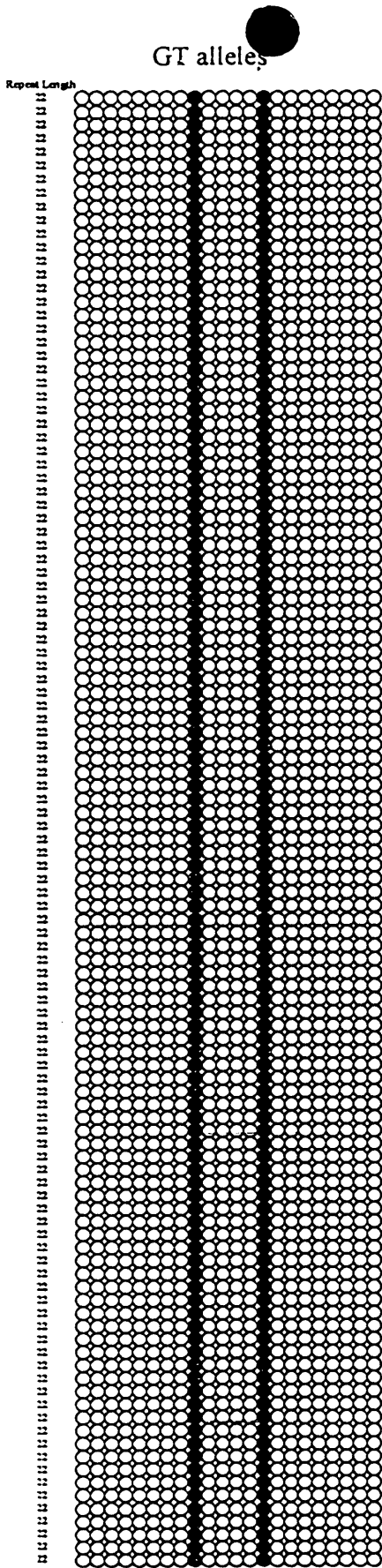


Figure 2

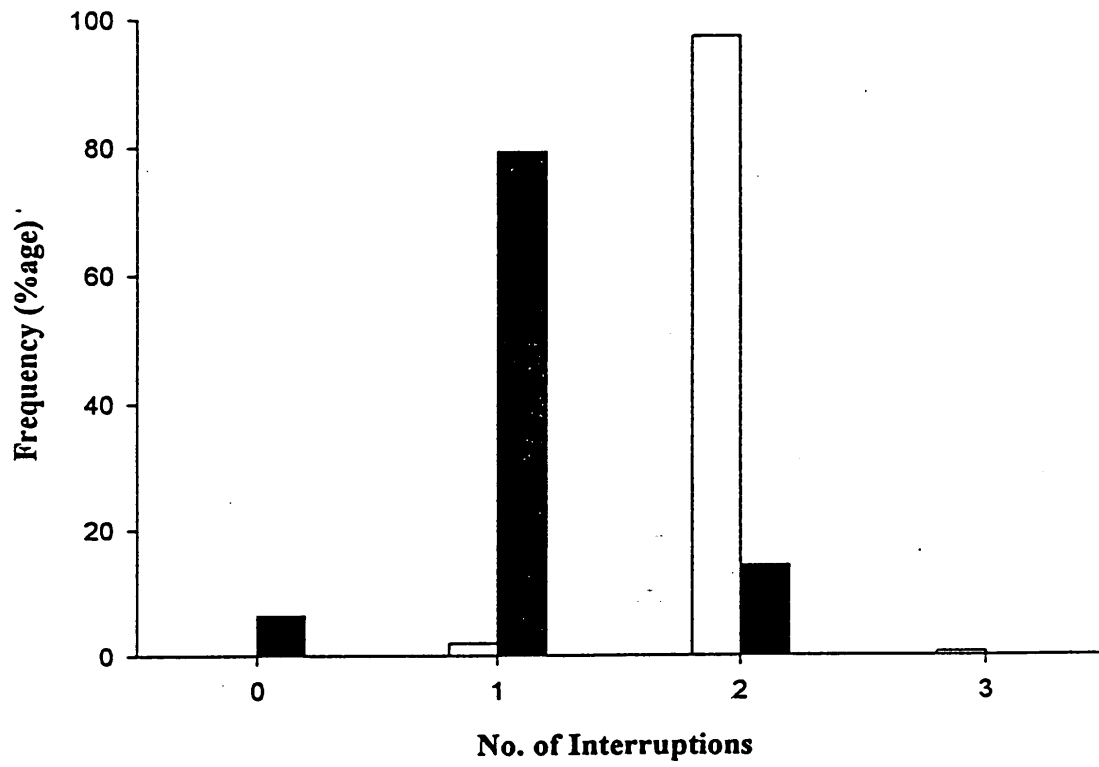
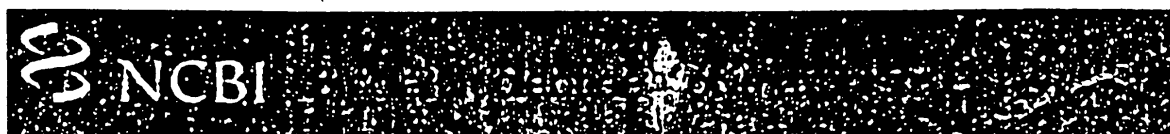


Figure 3



SNP Details

General

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SNP:

Handle|local_snp_id: FGU-CBT | SKB.2K.1.1

NCBI Assay Id(ss#): 869704

Reference SNP Id(rs#): 695871

STS Information: Not submitted

From SNP Database:

Submitter Handle: FGU-CBT
 Submitter Batch ID: SKB.2K.1
 Release Date: Aug 2 2000 2:53PM
 Molecular type: Genomic
 No. of Chromosomes sampled: 215
 Synonym defined:
 Organism: Homo sapiens
 Population: INDRPOP
 Submitter Method ID: SCA2-SNP
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

[View citation details](#)

NCBI Assay ID: 869704
 Submitter SNP ID: SKB.2K.1.1
 Synonyms:
 LOCUSID: 6111
 Submitter STS ID:
 STS Accession: not available
 GenBank Accession: U70121
 Gene Name: Human ataxin-2 gene/ Spinocerebellar ataxia 2 (SCA2) gene
 Length: 459

Flanking Sequence Information:

5' Assay: CTCGCGCTCA GACTGTTTTC GTAGCAACGG CAACGCGCGC GCGCGGTTTC GCGCGGCTC
 CCGCGGCTC CTTGGTCTCG GCGGCGCTCC CCGCGCTTC GTGCTC

Observed: G/C

3' Assay: TCCTTCTCCC CCTCGCCAGC CCGGGCGCCC CTCCGCGCGC GCCAACCCTC GCCTCCCCCG
 TCGCGGCCCC TCGTCCCCCG CCGCGTTCCG GCGTCTCTT GCGCGGCCCC GCTCCCGGCT
 GTCCCCCCCC GCGGTGCGAG CCGGTGTATG GCGCCCTCAC CATGTGCT

3' Flank: GAAGCCCCAG CAGCAGCAGC AGCAGCAGCA GCAACAGCAG CAGCAGCAAC AGCAGCAGCA
 GCAGCAGCAG CAGCCGCCCC CCGCGGCTCC CAATGTCCGC AAGCCCCGCG GCAGCGGCT
 TCTAGCGTGC CCGCGGCGCG CCGCTTCGCC GTCTCTGTC TCGTCTCTT CGTCTCGGC
 CAC

Allele Frequency Information:

POP Batch Id: SKB.2K.1:

Submitter Handle: FGU-CBT
 Submitter Method ID: SCA2-SNP
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

Handle|PopulationID: FGU-CBT| INDRPOP
 No. of Chromosomes Sampled: 215

Allele: C = 0.293 / G = 0.707

Fig.4(Cont.)



Reference SNP Record

NCBI SNP ID: rs695871

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NCBI Resource Links

GenBank: [U70323](#)
 LocusLink: no link established
 Integrated Maps: *under construction*

Submitter records for this ID:

Assay ID	Handle Local Submitter ID	Release Date
ss869704	FGU-CBT SKB.2K.1.1	Aug 2 2000 2:53PM

Variation Summary:

Assay sample size (number of chromosomes) : 430
 Population data sample size (number of chromosomes) :
 Total number of populations with frequency data: 1
 Total number of individuals with genotype data: 0
 Average estimated heterozygosity: 0.414
 Average Allele Frequency:

C:	0.293
G:	0.707

Validation Summary:

Marker displays Mendelian segregation: UNKNOWN
 PCR results confirmed in multiple reactions: YES
 Homozygotes detected in individual genotype data: UNKNOWN
 Insufficient genotype data to compute the goodness of fit to Hardy-Weinberg
 Insufficient data to compute individual x genotype consistency measures
 Validation status: *under construction*



Submitter Contact Details

General

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[Chromosome Report](#)

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 email: shwetachoudhry@hotmail.com
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 institution: Centre for Biochemical Technology (CSIR)
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.

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 phone: +91-11-7416489
 email: skb@cbt.res.in
 lab: Functional Genomics Unit
 institution: Centre for Biochemical Technology (CSIR),
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.

**Population**

SNP Population Details - 558

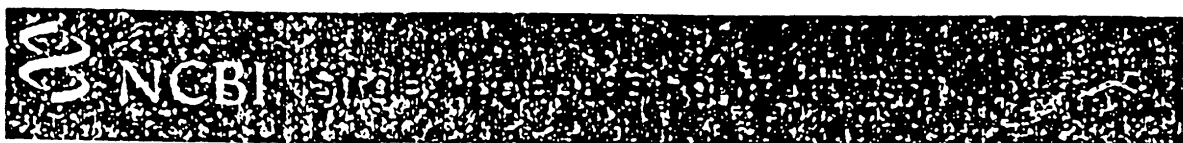
General[SNP Home](#)[dbSNP Summary](#)[How To Submit](#)[Genome SNP RFA](#)[FAQ](#)[RefSNP Summary Info](#)[FTP SERVER](#)[Database Schema](#)[Blast SNP](#)[Submission Form](#)

Submitter Population Handle: FGU-CBT
Submitter Population ID: INDPOP
Population Text:

Continent: Asia
Nation: India

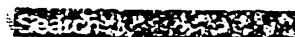
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Method

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Submitter Method Handle: FGU-CBT
 Submitter Method ID: SCA2-SNP
 Method Text:

The region containing the SNP was PCR amplified using the primers SCA2-FP3 (5' CTCCGCCTCAGACTGTTTTGGTAG 3') and SCA2-RP3 (5' GTGGCCGAGGACGAGGAGAC 3'). Approximately 100ng of genomic DNA was amplified in a 50 ml reaction volume containing a final concentration of 5mM Tris, 25mM KCl, 0.75mM MgCl₂, 0.05% gelatin, 20pmol of each primer and 0.5U of Taq DNA polymerase. Samples were denatured at 94°C for 3 min followed by 35 cycles of denaturation (94°C, 45sec), annealing (52°C, 30sec), extension (72°C, 45sec) and a final extension of 7 min at 72°C in a Perkin Elmer GeneAmp PCR System 9600. The PCR product was purified from band cut out of the agarose gel using QIAquick gel extraction kit (Qiagen) and was directly sequenced using dye terminator chemistry on an ABI Prism 377 automated DNA sequencer with the PCR primers.

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Publication

SNP Publication Details

[SNP Home](#)[dbSNP Summary](#)[How To Submit](#)[Genome SNP RFA](#)[FAQ](#)[RefSNP Summary Info](#)[FTP SERVER](#)[Database Schema](#)[Last SNP](#)[Submission Form](#)

Submitter Handle: FGU-CBT

pmid:

MEDUID:

TITLE:

Single Nucleotide Polymorphism in SCA2 Gene.

AUTHOR:

CHOUDHRY, S.; BRAHMACHARI, S.K.

JOURNAL:

VOLUME:

SUPPL:

ISSUE:

I_SUPPL:

PAGES:

YEAR: 2000

STATUS: 1- unpublished

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[Population](#)
[Publication](#)
[By Gene Name](#)
[Chromosome Report](#)

SNP:
 Handle|local_snp_id: FGU-CBT | SKB.2K.1.2
 NCBI Assay Id(ss#): 869705
 Reference SNP Id(rs#): 695872

STS Information: Not submitted

From SNP Database:

Submitter Handle: FGU-CBT
 Submitter Batch ID: SKB.2K.1
 Release Date: Aug 2 2000 2:53PM
 Molecular type: Genomic
 No. of Chromosomes sampled: 215
 Synonym defined:
 Organism: Homo sapiens
 Population: INDPOP
 Submitter Method ID: SCA2-SNP
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

[View citation details](#)

NCBI Assay ID: 869705
 Submitter SNP ID: SKB.2K.1.2
 Synonyms:
 LOCUSID: 6311
 Submitter STS ID:
 STS Accession: not available
 GenBank Accession: U70323
 Gene Name: Human ataxin-2 gene/ Spinocerebellar ataxia 2 (SCA2) gene
 Length: 459

Flanking Sequence Information:

5' Assay: CTCGCCTCA GACTGTTTG GTAGCAACGG CAACGGCGGC GCGCGTTTC GGCCCGGCTC
 CCGCGGGCTC CTTCGGTTCG GCGGGCTCC CCGCCCTTC GTCTCGTCC TTCTCCCCCT
 CGCCAGCCCG GCGCCCTTC CGGCCGCGC AACCCGCGC TCCCCGCTG GCGCCCG

Observed: T/C

3' Assay: GCGTCCCCGC CCGGTTCGCG CGTCTCCTTG GCGCGCCCGG CTCCCCGCTG TCCCCGCCCCG
 GCGTGCGAGC CGGTGTATGG GCCCCACAC ATGTGCT

3' Flank: GAAGCCCCAG CAGCAGCAGC AGCAGCAGCA GCAACAGCAG CAGCAGCAAC AGCAGCAGCA
 GCAGCAGCAG CAGCCGCGCG CCGCGGCTGC CAATGTCCGC AAGCCCGGCG GCAGCGGCCCT
 TCTAGCGTGC CCGCGCGCGG CGCCTTCGCC GTCTTCGTCC TCGGTCTCCT CGTCTTCGGC
 CAC

Allele Frequency Information:

POP Batch ID: SKB.2K.1:

Submitter Handle: FGU-CBT
 Submitter Method ID: SCA2-SNP
 Citation:
 Single Nucleotide Polymorphism in SCA2 Gene.

Handle|PopulationID: FGU-CBT|INDPOP
 No. of Chromosomes Sampled: 215

Allele: C = 0.293 / T = 0.707

RefSNP Record

Reference SNP Record

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[FAQ](#)
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[Database Schema](#)
[Blast SNP](#)
[Submission Form](#)

NCBI SNP ID: rs695872

NCBI Resource Links

GenBank: [U70323](#)

LocusLink: no link established

Integrated Maps: *under construction*

Submitter records for this ID:

Assay ID	Handle Local Submitter ID	Release Date
ss869705	FGU-CBT SKB.2K.1.2	Aug 2 2000 2:53PM

Variation Summary:

Assay sample size (number of chromosomes) : 430
Population data sample size (number of chromosomes) :
Total number of populations with frequency data: 1
Total number of individuals with genotype data: 0
Average estimated heterozygosity: 0.414
Average Allele Frequency:

C	0.293
T	0.707

Validation Summary:

Marker displays Mendelian segregation: UNKNOWN
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Homozygotes detected in individual genotype data: UNKNOWN
Insufficient genotype data to compute the goodness of fit to Hardy-Weinberg
Insufficient data to compute individual x genotype consistency measures
Validation status: *under construction*

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Publication

SNP Publication Details

[SNP Home](#)[dbSNP Summary](#)[How To Submit](#)[Genome SNP RFA](#)[FAQ](#)[RefSNP Summary Info](#)[FTP SERVER](#)[Database Schema](#)[Blast SNP](#)[Submission Form](#)

Submitter Handle: FGU-CBT

pmid:

MEDUID:

TITLE:

Single Nucleotide Polymorphism in SCA2 Gene.

AUTHOR:

CHOUDHRY,S.; BRAHMACHARI,S.K.

JOURNAL:

VOLUME:

SUPPL:

ISSUE:

I_SUPPL:

PAGES:

YEAR:

2000

STATUS:

1- unpublished

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Submitter Population Handle: FGU-CBT
Submitter Population ID: INDPOP
Population Text:

Continent: Asia
Nation: India

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009071 61620 07919 110800

Method

SNP Method Details - 564

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Submitter Method Handle: FGU-CBT
 Submitter Method ID: SCA2-SNP
 Method Text:

The region containing the SNP was PCR amplified using the primers SCA2-FP3 (5' CTCGCCCTCAGACTGTTTGGTAG 3') and SCA2-RP3 (5' GTGCCCGAGGACGAGGAGAC 3'). Approximately 100ng of genomic DNA was amplified in a 50 ml reaction volume containing a final concentration of 5mM Tris, 25mM KCl, 0.75mM MgCl₂, 0.05% gelatin, 20pmol of each primer and 0.5U of Taq DNA polymerase. Samples were denatured at 94°C for 3 min followed by 35 cycles of denaturation (94°C, 45sec), annealing (52°C, 30sec), extension (72°C, 45sec) and a final extension of 7 min at 72°C in a Perkin Elmer GeneAmp PCR System 9600. The PCR product was purified from band cut out of the agarose gel using QIAquick gel extraction kit (Qiagen) and was directly sequenced using dye terminator chemistry on an ABI Prism 377 automated DNA sequencer with the PCR primers.

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Submitter

Submitter Contact Details

[SNP Home](#)[dbSNP Summary](#)[How To Submit](#)[Genome SNP REA](#)[FAQ](#)[RefSNP Summary Info](#)[FTP SERVER](#)[Database Schema](#)[Blast SNP](#)[Submission Form](#)[Main Search](#)[By Submitter](#)[New Batches](#)[Method](#)[Population](#)[Publication](#)[By Gene Name](#)[Chromosome Report](#)

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 email: skb@cbt.res.in
 lab: Functional Genomics Unit
 institution: Centre for Biochemical Technology (CSIR),
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.



Nucleotide

Search for

Limits Index History Clipboard

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☐ 1: GI = "1679683" [GenBank] Human ataxin-2 (SCA2) mRNA... PubMed, Protein, Related Sequences, Taxonomy, OMIM, LinkOut

LOCUS HSU70323 4481 bp mRNA PRI 20-NOV-1996

DEFINITION Human ataxin-2 (SCA2) mRNA, complete cds.

ACCESSION U70323

VERSION U70323.1 GI:1679683

KEYWORDS

SOURCE human.

ORGANISM *Homo sapiens*
Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.

REFERENCE 1 (bases 1 to 4481)
AUTHORS Pulst, S.-M., Nechiporuk, A., Nechiporuk, T., Gispert, S., Chen, X.-N.,
Lopes-Cendes, I., Pearlman, S., Starkman, S., Orozco-Diaz, G.,
Lunkes, A., DeJong, P., Rouleau, G.A., Auburger, G., Korenberg, J.R.,
Figueroa, C. and Sahba, S.
TITLE Moderate expansion of a normally biallelic trinucleotide repeat in
spinocerebellar ataxia type 2
JOURNAL Nature Genet. 14 (3), 269-276 (1996)

MEDLINE 97051920
REFERENCE 2 (bases 1 to 4481)
AUTHORS Pulst, S.-M.
TITLE Direct Submission
JOURNAL Submitted (10-SEP-1996) Medicine, Cedars-Sinai, 8700 Beverly Blvd.,
Los Angeles, CA 90048, USA

FEATURES
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Figure : 6

NCBI Sequence Viewer

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1981	gtctctacta	tgcttaaacg	catgtcttca	gaaggggctc	caaggaatgc	ccaaagggcg
2041	cagcgacatc	ctcgaaatca	cagagtttct	gctggggagg	gtttcatatc	cagtgggcta
2101	gaatttggat	ccccacaacc	accagatgaa	gcagcttact	ctccagtagc	aaagccaggt
2161	ccctcggggg	gaacgttggt	atcagtggtc	agtggggttc	ccaagttatc	ccctaaaaat
2221	catagaccac	ggtctcccg	acagaacagt	attggaaata	ccccagtg	ggcagttctt
2281	gtctctcccc	aaagctggat	tatttccaat	gaagctgttg	ccatgcctat	tcacagctga
2341	tctctctacg	ctcgtagtcc	tgcatgaaac	agagctgtta	ccctcttag	tgaagctaaa
2401	gattccagcg	ttcaagatca	gaggcagaac	tctcttgca	ggaataaaga	aaatattaaa
2461	cccaatgaaa	catcacctag	ctcttcaaaa	gtgaaaaaca	aggatataat	accagttgtt
2521	ctgaaacata	gaaaaacagt	tgatgattta	aagaaataat	aaagtatatt	taggttacag
2581	ccaagtctta	cttctgaaac	tatggattca	ctacttaaaa	aaaaatagaa	gggagaaaaa
2641	tcaagagatt	tgatcaaaag	caaaaatgaa	ccaagtgcta	aggattcttt	catgaaaaat
2701	acgacgagca	actgtaccag	tgggcagcag	aagcggaaat	ggccccagat	ttccccctca
2761	ataacttagta	acacggagca	caaaagggga	cctgaggtca	cttcccaagg	gggttcagact
2821	tccagcccgag	catgttaaaa	agagaaagac	gataaaggag	cgtaaaagaa	cgacgttagc
2881	caagttagga	aatcaacatt	gaatcccaat	gcaaaaggag	tcaacccacg	ttccttctct
2941	cagccaacagc	cttctactac	cccaacttca	ctctggccctc	aagcacaacc	tagcccatct
3001	atggtgggtc	atcaaacgac	aaactccagtt	tatactcagc	gtgttggtt	tgcaaccaat
3061	atgatgtatc	cagtcctcag	gaagccagac	gtgcacactc	tatacccaat	acctatgacg
3121	cccatgccag	tgaaatcaag	cagacataat	agagcagtat	caaatatgcc	ccaaacaggg
3181	caagacacagc	atcatcagag	tgccatgatg	caccacagct	gcagcagggg	cccaccgatc
3241	gcagcaccccc	caccagctta	cttcgcgcga	tatgttgctc	acagtcctca	gcagttccca
3301	aaacagcccc	ttgttcagca	ttgtgccaat	tactcagctc	ctccacttca	tgcttatagc
3361	cctgtaaatc	agggtaaatg	tag			

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Fig.6(Cont.)